Course Specifications
Valid as from the academic year 2018-2019

Advanced Human Genetics (D012701)

Course

Course offerings in academic year 2018-2019
A (semester 2) English

Lecturers in academic year 2018-2019
Coucke, Paul GE02 lecturer-in-charge
Symoens, Sofie GE02 co-lecturer
Vanacker, Olivier GE02 co-lecturer
Van Coster, Rudy GE02 co-lecturer

Offered in the following programmes in 2018-2019
Master of Science in Biomedical Sciences crdts offering
6 A

Teaching languages
English

Keywords
Epigenetics, imprinting, uniparental disomy, haplo-insufficiency, dominant negative, gain-of-function, triplet repeat, genomic disorders, single nucleotide polymorfisms, non-mendelian inheritance, farmacogenetics, gene-therapy

Position of the course
In this course we will focus on new insights in the genetic basis of human disease. In addition to classic nucleotide changes in the coding region of genes, positional and epigenetic effects gain increasing importance. This course discusses state-of-the-art research papers that will provide recent insights in new genetic mechanisms at the basis of human disease. Other topics include new molecular genetic analysis techniques, recent insights in the pathogenetic mechanisms and the therapeutic applications. Special attention is paid to the role of genetics in the development of new therapeutic strategies. Students will gain insight in diverse aspects of human genetics of the 21st century.

Contents
The following topics will be covered:
1. Epigenetics and imprinting
2. DNA methylation en histonacetylation
3. MicroRNA and other non protein coding RNAs
4. Disease mechanisms (haplo-insufficiency, dominant negative, gain-of-function, triplet repeat)
5. Genomic disorders (recurrent deletions)
6. Long range regulation of transcription, conserved non-genic sequences
7. Non-mendelian inheritance/pedigree analysis
8. Complex inheritance
9. Mitochondrial genetics
10. Pharmacogenetics
11. Gene therapy

Initial competences
Having successfully completed the courses Human genetics, Molecular biology I, Molecular Biology II and Human pathogenesis from the bachelor program biomedical sciences, or having acquired the relevant ending objectives by other means.

Having completed successfully the bachelor degree in biomedical sciences or having

(Course size (nominal values; actual values may depend on programme)
Credits 6.0 Study time 180 h Contact hrs 55.0 h

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(Approved)
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Final competences
1. To gain new insights in the most actual developments of gene discovery, pathogenetic mechanisms and therapeutic strategies of human genetic conditions.
2. To be able to critically read and discuss scientific papers concerning gene discovery, pathogenic mechanisms and therapeutic strategies.

Conditions for credit contract
Access to this course unit via a credit contract is determined after successful competences assessment

Conditions for exam contract
This course unit cannot be taken via an exam contract

Teaching methods
Lecture, microteaching

Extra information on the teaching methods
Group discussions, lectures, debates

Learning materials and price
Dutch syllabus, completed with scientific papers (English) and slides available via Minerva.

References
• State-of-the-art articles

Course content-related study coaching
Interactive support via questions and answers during lectures and by email and Minerva.

Evaluation methods
End-of-term evaluation

Examination methods in case of periodic evaluation during the first examination period
Written examination

Examination methods in case of periodic evaluation during the second examination period
Written examination

Examination methods in case of permanent evaluation
Oral examination

Possibilities of retake in case of permanent evaluation
Examination during the second examination period is possible in modified form

Calculation of the examination mark

(Approved)